The HCU Herald

More HCU News Inside

HCU HERO:
GARRETT FROM ILLINOIS

How were you diagnosed?

My name is Garrett, and I was diagnosed with Homocystinuria when I was 5 years old. I was playing on my toy rocking boat and managed to build enough momentum to move the boat backwards enough that I ended up smacking the back of my head on a cabinet, which knocked me unconscious.

From what I'm told, I was rushed to the hospital. While some things got better, I noticed that there was something not right with my eyes and they didn't seem to be getting better, even with the medicine they prescribed. I believe they originally thought I had pink eye, but later decided that it had to be one of two things. They quickly disqualified me for one of the ideas they had and were sure it was Homocystinuria. It was also discovered that my lenses had detached, and if I didn't get them removed immediately, I was going to go blind.

So, there's my origin story… I wanted it to be a radioactive spider that bit me, but not so, unfortunately.

How has HCU affected you, your family and relationships?

HCU has affected me in many ways, but probably not really any different from most patients with Homocystinuria. Family parties are a constant drag when it comes to food. There are always things present that I cannot eat, but people will still ask if I can eat them even if they know I can't. I've had to learn to bring my own food, because most of the time all that is available that I can eat is salad or fruit – not exactly filling.

What are some of your successes with HCU?

Growing up I was told by numerous teachers that I wouldn't go to college, and in some cases was told I would drop out of high school. They would accuse me of not trying or not working hard enough. In reality, I would stay up until midnight trying to complete my assignments. I even had extra tutors to help me. I realized I had two choices: I could let them win or I could defy their expectations. While I've long been graduated from high school and am still in college, I don't let this get me down. I've gotten my associates degree, and I am about to transfer to Northern Illinois University (NIU) to pursue my passion of photography as my career. You can follow my artwork on instagram at:
https://www.instagram.com/wadesworldofphotography/

What are some of the challenges you have faced?

Cost. I'm sure many with HCU are aware of how expensive low protein medical foods are. Luckily with time, there are more low protein food companies, and food choices with a little better prices. Right now my "go to" choice for staples like bread, pasta, and rice is Flavis. It seems to be the most affordable option to my knowledge. I suggest if your insurance will cover low protein food you use it. Being able to piggy back off the Gluten-free, Dairy-free and Vegan community, when possible, has made finding foods much easier than it was 10 years ago.

Eating out is a constant struggle, but has gotten better. A man can only eat so many potatoes and salads before he questions his life choices, but on a serious note food at restaurants has gotten better (big shout out to the Vegan and Gluten-free communities as my diet would be a lot worse if it wasn't for you). There are some restaurants like Noodles and Company that carry vegetable and gluten-free noodles. Other places like Mod Pizza carry a gluten-free crust and vegan cheese. Some restaurants are willing to cook the medical food even if you ask. What I'm trying to say is don't be afraid to ask and see what your options are for times have changed. There's a lot more availability now and people are willing usually to work with you if you explain the situation.

What words of advice do you have to newly diagnosed patients and families?

Words of advice I have for anyone newly diagnosed – be the best advocate for yourself that you can be. No one else knows you the way you do and ultimately you are the one responsible for yourself – not anyone else.

Get involved in the community as much as possible. This is something that no one should have to deal with alone. Ask others what works for them, whether it be recipes, different forms of medication, insurance issues, or how to go about working with others outside of your community like at school or work. I remember a former doctor I had was not very encouraging to me and my sister in connecting us with others. Reach out to others; if it wasn't for them our quality of life wouldn't be as good as it is. Last word of advice – do not let HCU define who you are or make you feel like a victim. You cannot rise above HCU or any other problem in life by letting yourself be a victim. You have to have the mentality of victory to rise above it as you do with all problems!

HCU COMMUNITY COOKBOOK

Amber's Kitchen

Basic Risotto
Serving Size: 1/3 Cup
Protein per Serving: 0.5 g
Calories per Serving: 177

Marsala Mushrooms
Serving Size: 30 g
Protein per Serving: 1.1 g
Calories per Serving: 254

Banana Zucchini Cake
Serving Size: 1 slice
Protein per Serving: 1 g
Calories per Serving: 197

Recipes at:
https://hcunetworkamerica.org/hcu-community-cook-book/

Rare Bears for Kids with HCU

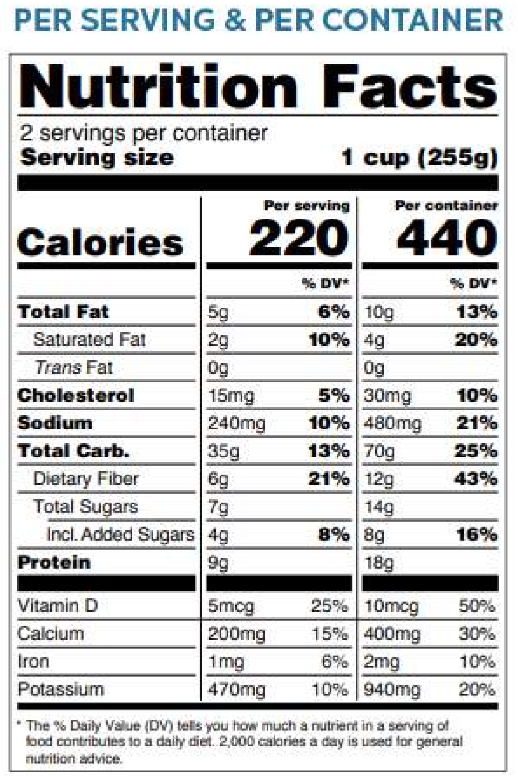
RARE science
Accelerating Cures for RARE Kids

We are thrilled to announce that the Rare Bears for HCU Campaign in partnership with Rare Science has been re-opened!

To enroll in the RARE Bear Program and to request a RARE Bear, please click the link and complete the form: https://www.rarescience.org/hcu/

- Those who have already received a bear, are not eligible
- Date for gifting will be announced later
- You will not receive a confirmation email or be notified when your bear has been shipped

FOOD NUTRITION LABEL CHANGES



PER SERVING & PER CONTAINER

Nutrition Facts

2 servings per container

Serving size **1 cup (255g)**

	Per serving		Per container	
Calories	**220**		**440**	
		% DV*		**% DV***
Total Fat	5g	**6%**	10g	**13%**
Saturated Fat	2g	**10%**	4g	**20%**
Trans Fat	0g		0g	
Cholesterol	15mg	**5%**	30mg	**10%**
Sodium	240mg	**10%**	480mg	**21%**
Total Carb.	35g	**13%**	70g	**25%**
Dietary Fiber	6g	**21%**	12g	**43%**
Total Sugars	7g		14g	
Incl. Added Sugars	4g	**8%**	8g	**16%**
Protein	9g		18g	
Vitamin D	5mcg	25%	10mcg	50%
Calcium	200mg	15%	400mg	30%
Iron	1mg	6%	2mg	10%
Potassium	470mg	10%	940mg	20%

* The % Daily Value (DV) tells you how much a nutrient in a serving of food contributes to a daily diet. 2,000 calories a day is used for general nutrition advice.

Many of you have noticed changes to the nutrition facts panel on the back of your favorite products. This is due to formatting changes mandated by the FDA. While they were authorized in 2016, smaller manufacturers had until January 2021 to publish the changes.

So what's going on?

The most dramatic changes are on those products that show both the per serving, and per container, nutritionals. Don't let that second column scare you -- focus on your per-serving protein content, and the serving size, and you should be fine.

Click through to read about the changes in more detail.

SCHOLARSHIP OPPORTUNITIES

Living with a rare disease means managing unique challenges, including frequent doctor visits, rigorous treatment regimens and hospitalizations, and exposure risks. While quality and duration of life continues to improve thanks to improved diagnosis and treatment approaches, individuals living with rare diseases still face disparities in achieving traditional life milestones.

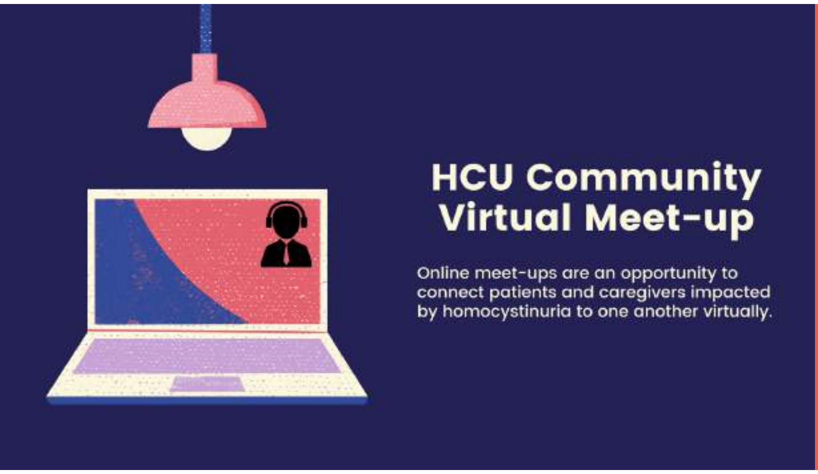
That's why The EveryLife Foundation for Rare Diseases established the #RAREis Scholarship Fund – to enrich the lives of adults living with rare diseases by providing support for their educational pursuits. Thanks to the support of Horizon Therapeutics RAREis, one-time awards of $5,000 each will be granted to up to 32 recipients for the Fall 2021 semester.

Deadline for Fall Semester 2021application is May 7, 2021 at 3:00 p.m. CDT Learn more and apply here: https://everylifefoundation.org/rare-scholarship/

UPCOMING EVENTS

Come check out our Virtual Homocystinuria Meet-ups!
Join our virtual meet-up for a chance to meet, connect, and learn from other patients and caregivers who are facing similar challenges. Whether it's navigating adherence issues, insurance, clinic visit, or life transitions, you are not alone.

Note our new registration page: https://hcunetworkamerica.org/virtual-meet-ups/



- **Sun., March 7 @ 7 pm CST**
- **Mon., March 29 th @ 1 pm CST | 7 pm UTC**

- **Sun., March 21 @ 7 pm CST**

2021 Land of the Free, Home of the Brave patient-family experince conference, POSTPONED!

Due to the ongoing pandemic HCU Network America, the Organic Acidemia Association, and the Propionic Acidemia Association have decided to to postpone their 2021 combined conference.

We will be sending out an email shortly with more information on future dates and some virtual opportunities that will take place this summer.

Save the Date for these Upcoming Events!

- 2021 Recordati HCU Click Campaign
- Race for Research, Virtual Race – September 1-30, 2021
- HCU Awareness Month – October 1-31, 2021
- Giving Tuesday – November 30, 2021

UPCOMING EVENTS

Registration for the RARE on the Road – Rare Disease Leadership Interactive Webinar is now OPEN!

On March 23, 2021 from 11:00am – 1:30pm ET, engage in interactive tutorials on how to tell your rare story and how to get involved in advocacy. This portion of the event will feature special guest speakers, including a rare disease patient and policy expert, there to provide insights to amplify the patient voice from anywhere in the U.S.

Whether you're new to the rare disease community or a "seasoned veteran," the RARE on the Road Rare Disease Leadership Interactive Webinar is for you! Click here to register for the interactive webinar.

If you're interested in attending one of the RARE on the Road State-Specific Training and Networking Sessions, save the dates and stay tuned for more details in the coming weeks. For more information, please visit Raretour.org.

May 4: Virtual State-Specific Training and Networking Event – Nevada
May 11: Virtual State-Specific Training and Networking Event – Florida
May 18: Virtual State-Specific Training and Networking Event – Illinois

Will you be the next HCU Patient Hero?

Email us your patient story at: dbartke@hcunetworkamerica.org

FEBRUARY FUNDRAISING ALL STARS!

Super Bowl Square Highlights

Big sporting events are a great way to not just show your team spirit, but they are a fantastic way to raise funds for your charity of choice - HCU Network America! Cole Sullivan did just that with the help of Danae' Bartke, Tom Hawkins, Kristin Rapp and Annette Settle during the Super Bowl LV game!They did not just fill up one squares board, but two and raised over $2,400 in the process.

Facebook Birthdays Fundraisers

We'd like to wish Ashlee W., Aimee W., Rebecca S., and Kara B. a HAPPY BIRTHDAY and thank them for choosing to dedicate their birthday fundraisers to HCU Network America and the Homocystinuria community! Their birthday fundraisers combined raised $1,190 for HCU Network America – unless otherwise designated, their funds will go towards our vital Homocystinuria education resources, programs and events! These resources provide the much needed support and understanding for patients, caregivers and medical professionals affected by Homocystinuria.

Rare Disease Day Fundraisers

If you are looking for easy way to bring awareness to Homocystinuria and to raise funds, setting up a Rare Disease Day fundraiser is just that! Jessica K. took advantage of Rare Disease Day, February 28th and set up a fundraiser to benefit HCU Network America. Jessica was able to raise $225! Way to go Jessica for taking the lead this Rare Disease Day!

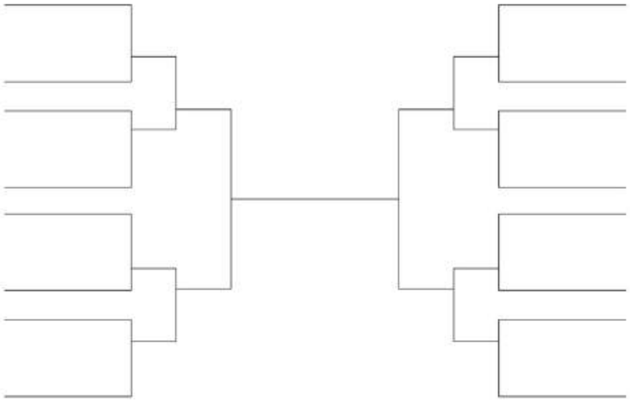
RARE DISEASE DAY®

Together this amazing group of Fundraising All-Stars raised over $3,800!

FEBRUARY FUNDRAISING EVENT: MARCH MADNESS

March Madness Selection Sunday is March 14! Start assembling your pool and **raise funding for the resources and tools HCU Network America!**

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!



How Do Madness Squares Work?

If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown

Each round is worth a set number of points. You can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner

Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

- For further instructions and to print your bracket, visit: https://www.printyourbrackets.com/howtomarchmadness.html
- For online tools, check out: https://www.runyourpool.com/march-madness-squares-pools.cfm

NEED HELP HOSTING A FUNDRAISER? EMAIL US - INFO@HCUNETWORKAMERICA.ORG

NEWS YOU SHOULD KNOW

New Cobalamin Steering Committee Member

My name is Victoria Lisa, I am 20 years old and attending Saint Peter's University, majoring in Criminal Justice. I live in Hoboken, New Jersey with my mom Carmella, my dad Pete, and my brother Peter, who is 23 years old and has CblC. I enjoy watching YouTube videos, exploring, dancing, and trying new things. Peter is always happy, always smiling. People in town know us because of Peter most of the time. He enjoys listening to all types of music; he can go from listening to rock and roll to listening to Barney.

Compassion Works Medical Launches Website

Many in our community have become quite familiar with Raenette Franco, the founder of Compassion Works Medical LLC. We are excited to share, that after 8 years of serving the metabolic community and helping them achieve medical foods coverage she has launched a website! Take a look!

GET INVOLVED:
NEWBORN SCREENING NEEDS YOU!

Newborn Screening Saves Lives Act Reauthorization

The Newborn Screening Saves Life Reauthorization Act has been introduced in the House. Please ask your Representative and Senators to support this essential bill that reauthorizes "critical federal programs that provide assistance to states to improve and expand their newborn screening programs, support parent and provider education, and ensure laboratory quality and surveillance for newborn screening."

Click here to take action on NBS today

Newborn Screening Survey - Have you shared it with your clinic?

WE HAVE AN OPPORTUNITY TO HELP CHANGE THE PROCESS BUT NEED YOUR STORY TO GIVE US THE EVIDENCE TO BUILD OUR CASE

But we have newborn screening for HCU...

According to recent statistics, approximately 25-50% of patients are missed by newborn screening for Classical Homocystinuria. There are multiple factors that can play into these numbers. Currently it is federal mandate that all states screen for Classical Homocystinuria through the newborn screening test, but there are no set standards. Meaning, every state or region can set their own methionine cut offs. A handful of states also do tier two testing—meaning they have a second round of newborn screening, making it more likely for homocystinuria to be picked up. Another factor that plays into the effectiveness of the test, is how elevated the patient's levels are at the time of the test. Patients who are pyridoxine (B6) responsive, or have more functioning CBS enzyme, are less likely to be picked up by the newborn screening.

So how can you help?

1) If you or your loved one were missed at screening, we need to hear from you ASAP so we have enough evidence to bring about change. Contact Danae (dbartke@hcunetworkamerica.org) if you can help us, and she will lead you through the process that is outlined below.

2) Talk to your geneticist about the newborn screening survey and urge them to complete it! This will help us build support for changes to the process to increase the likelihood that HCU patients will be diagnosed at birth.

Here is the letter portion we would ask you to give to your clinic, followed by the survey form:

GET INVOLVED:
NEWBORN SCREENING NEEDS YOU!

To Whom this may concern,

I would appreciate your support in answering a brief survey to help support efforts to improve newborn screening for classical homocystinuria (HCU).

I have been working with HCU Network America, a patient advocacy and support group for HCU, for whom I serve as a medical advisor. One of their key goals is to improve newborn screening for HCU, as it is estimated that over half of patients are missed by the current screening process and often are not diagnosed until they have developed serious clinical symptoms. To build support for an improved process, we are collecting information on patients missed by the current screening process, which we intend to then publish in a consolidated case report.

Please support our efforts by completing the attached brief questionnaire at https://hcunetworkamerica.org/survey-on-classical-homocystinuria-patients-missed-by-newborn-screening/

Sincerely,
Can Ficicioglu, M.D., Ph. D.
Director of Newborn Metabolic Screening Program, Children's Hospital of Philadelphia

Survey on Classical Homocystinuria (HCU) Patients Missed by Newborn Screening

Do you have any patients with classical HCU missed by NBS and diagnosed later based on symptoms?
( ) Yes ( ) No

If yes, at what age were the patients diagnosed, and what year were they born and in what state?

Age at diagnosis (mos.) ____ Year of birth _____ State born___________
Age at diagnosis (mos.) ____ Year of birth _____ State born___________
Age at diagnosis (mos.) ____ Year of birth _____ State born___________

Would you be willing to provide information to contribute to a "Case Report" we plan to publish on patients missed by Newborn Screening?

What is the name and address of your clinic and the best contact person for further information?
Clinic Name
Clinic address
Contact Person:

- Name
- E-mail
- Phone

Please send completed survey to Dr. Can Ficicioglu at ficicioglu@email.chop.edu

Live better, together!

Contact Register

What is the contact register?

The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission. By registering, you will be able to identify other patients in your state and request their contact information. You will also be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?

HCU Network America strives to inform patients and families with resources, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission - plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://hcunetworkamerica.org/contact-register/

FOLLOW US